

METHODS OF DNA MARKER-BASED GENETIC ANALYSIS USING ESTIMATED HAPLOTYPE FREQUENCIES AND USES THEREOF

Abstract

5 The present invention is primarily drawn to methods of DNA marker-based genetic analysis
using estimated haplotype frequencies to draw inferences about the relationship between haplotypes
and traits or diseases. Unlike many haplotype analysis methods that require phase information that
can be difficult to obtain from samples of non-haploid species, the instant methods are based on
strategies for estimating haplotype frequencies from unphased diploid genotype data using the
10 Estimation-Maximization (E-M) algorithm to overcome the missing phase information. These
estimated haplotype frequencies can then be used in a variety of statistical analyses, including those
to infer the existence of a disease gene. The process can include: 1) estimating haplotype
frequencies; 2) computing test statistics; and 3) drawing inferences.

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